

MAN2B1 gene

mannosidase alpha class 2B member 1

Normal Function

The *MAN2B1* gene provides instructions for making the enzyme alpha-mannosidase. This enzyme works in the lysosomes, which are compartments that digest and recycle materials in the cell. Within lysosomes, the enzyme helps break down complexes of sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins). In particular, alpha-mannosidase helps break down oligosaccharides containing a sugar molecule called mannose.

Health Conditions Related to Genetic Changes

Alpha-mannosidosis

More than 120 mutations in the *MAN2B1* gene have been identified in people with alpha-mannosidosis, a rare inherited disorder that causes problems in many organs and tissues of the body. Affected individuals may have intellectual disability, distinctive facial features, and skeletal abnormalities. Some of the *MAN2B1* gene mutations that cause alpha-mannosidosis change one protein building block (amino acid) in the alpha-mannosidase enzyme. Other mutations result in an abnormally shortened enzyme, or cause the enzyme to be pieced together incorrectly.

These mutations interfere with the ability of the alpha-mannosidase enzyme to perform its role in breaking down mannose-containing oligosaccharides. These oligosaccharides accumulate in the lysosomes and cause the cells to malfunction and eventually die. Tissues and organs are damaged by the abnormal accumulation of oligosaccharides and the resulting cell death, leading to the characteristic features of alpha-mannosidosis.

Other Names for This Gene

- LAMAN
- lysosomal acid alpha-mannosidase
- MA2B1_HUMAN
- MANB
- mannosidase, alpha B, lysosomal

- mannosidase, alpha, class 2B, member 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MAN2B1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4125\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4125[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28MAN2B1%5BTIAB%5D%29+OR+%28%28LAMAN%5BTIAB%5D%29+OR+%28lysosomal+acid+alpha-mannosidase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- MANNOSIDASE, ALPHA, CLASS 2B, MEMBER 1 (<https://omim.org/entry/609458>)

Research Resources

- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MAN2B1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MAN2B1[gene]))
- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4125>)

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Genomic Location

The *MAN2B1* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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